FACT SHEET
Healthcare Provider

Congenital Hypothyroidism (CH)

Description:
Congenital Hypothyroidism (CH) is a condition in which the body does not secrete sufficient amounts of thyroid hormone**.** Because the body cannot produce an adequate amount of thyroid hormone, the pituitary will make additional thyroid stimulating hormone (TSH) in an attempt to entice the thyroid to produce more hormone. The causes for the lack of hormone production can be from agenesis, ectopic thyroid gland, or inherited disorders of thyroid hormone biosynthesis. If untreated, the symptoms of hypothyroidism will usually progress. Thyroxine deficiency in infancy can cause severe, irreversible mental and physical retardation, a condition known as cretinism. CH occurs sporadically and is not usually an inherited disorder. This disorder is not associated with any prenatal lifestyle or risk factors.

Incidence in General Population:
- 1:5,000 live births
- The occurrence is higher in the Hispanic and Native American ethnic groups. It is twice as common in females as in males. CH is more common in Caucasians than African Americans.

Symptoms:
- Feeding problems
- Lethargy
- Prolonged postnatal jaundice
- Delayed stooling and constipation
- Enlarged protruding tongue
- Course hair
- Cold intolerance
- Protruding abdomen
- Umbilical hernia
- Cold mottled skin
- Irritability
- Sluggish reflexes
- Patent posterior fontanelle
- Widely spread cranial sutures or delayed skeletal maturation for gestational age

Newborn Screening Technology:
Detection through an immunofluorescent assay (IFA) for thyroxine (T4) and thyroid stimulating hormones (TSH). IFA first measures the level of T4 in the blood. For infants whose T4 level falls in the lowest 10% of the results for the assay, TSH is measured on the same specimen. An elevated level of TSH indicates primary hypothyroidism, and the responsible physician is directed to have confirmatory T4 and TSH tests performed on a sample of the infant's serum.

Diagnosis:
Collection of serum TSH and T4 levels is recommended along with referring infants to a pediatric endocrinologist for evaluation.

Monitoring:
Growth and development must be monitored at frequent intervals, including measurement of thyroid hormone levels to prevent both under and over treatment and their associated morbidities.

Treatment:
Early and adequate treatment and regular, careful monitoring is important to prevent permanent retardation of intellectual function and/or skeletal growth. With early treatment neurological development is comparable to peers without this diagnosis. Treatment for this disorder is lifelong. Medications should be prescribed and followed closely by a pediatric endocrinologist. Levothyroxine is given orally at a dosage to produce a T4 concentration in the upper normal range to normalize TSH levels. Tablets should be crushed daily; mixed with a few milliliters of water, formula, or breast milk; and fed to infant. Levothyroxine should not be mixed with soy formula or with formula containing iron, as these products interfere with absorption of the medication. Dosages of medication will need to be gradually increased as the infant grows.

Immunizations:
Immunization schedules should be followed to ensure protection from all other childhood diseases.

Growth and Development:
It is crucial to monitor all growth parameters on a regular basis.

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